



## GNAI3 gene

G protein subunit alpha i3

### Normal Function

The *GNAI3* gene provides instructions for making one component, the inhibitory alpha subunit, of a protein complex called a guanine nucleotide-binding protein (G protein). G proteins are composed of three protein subunits: alpha, beta, and gamma. Each of these subunits is produced from a different gene.

Through a process called signal transduction, G proteins trigger a complex network of signaling pathways within cells. These pathways help transmit information from outside the cell to inside the cell. Specifically, G proteins made with the *GNAI3* inhibitory alpha subunit reduce (inhibit) the activity of an enzyme called adenylyl cyclase, which is an important chemical messenger within cells. G protein signaling ultimately influences many cell activities, instructing the cell to grow, divide, or take on specialized functions.

Studies suggest that G protein signaling involving the *GNAI3* inhibitory alpha subunit contributes to the development of the first and second pharyngeal arches. These embryonic structures ultimately develop into the jawbones, facial muscles, middle ear bones, ear canals, outer ears, and related tissues.

### Health Conditions Related to Genetic Changes

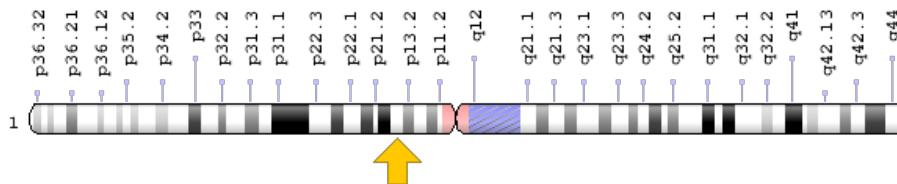
#### auriculo-condylar syndrome

At least two mutations in the *GNAI3* gene have been found to cause auriculo-condylar syndrome, a disorder that primarily affects the development of the ears and lower jaw (mandible). The identified mutations change single protein building blocks (amino acids) in the inhibitory alpha subunit. These mutations likely alter the structure of the inhibitory alpha subunit and impair G protein signaling. Abnormal signaling alters the formation of the lower jaw: instead of developing normally, the lower jaw becomes shaped more like the smaller upper jaw (maxilla). The abnormal shape leads to an unusually small chin (micrognathia) and problems with jaw function. Researchers are working to determine how mutations in this gene lead to the other developmental abnormalities associated with auriculo-condylar syndrome.

## Chromosomal Location

Cytogenetic Location: 1p13.3, which is the short (p) arm of chromosome 1 at position 13.3

Molecular Location: base pairs 109,548,564 to 109,595,843 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- 87U6
- ARCND1
- g(i) alpha-3
- GNAI3\_HUMAN
- guanine nucleotide binding protein (G protein), alpha inhibiting activity polypeptide 3
- guanine nucleotide-binding protein G(k) subunit alpha

## Additional Information & Resources

### Educational Resources

- Basic Neurochemistry (sixth edition, 1999): Adenylyl Cyclases  
<https://www.ncbi.nlm.nih.gov/books/NBK27958/>
- Molecular Biology of the Cell (fourth edition, 2002): Signaling through G-Protein-Linked Cell-Surface Receptors  
<https://www.ncbi.nlm.nih.gov/books/NBK26912/>

## Scientific Articles on PubMed

- PubMed

<https://www.ncbi.nlm.nih.gov/pubmed?term=%28GNAI3%5BTIAB%5D%29+OR+%28guanine+nucleotide+binding+protein%5BTIAB%5D%29+AND+%28alpha+pha+3%5BTIAB%5D%29%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+NOT+%28achromatopsia%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>

## OMIM

- GUANINE NUCLEOTIDE-BINDING PROTEIN, ALPHA-INHIBITING ACTIVITY POLYPEPTIDE 3  
<http://omim.org/entry/139370>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_GNAI3.html](http://atlasgeneticsoncology.org/Genes/GC_GNAI3.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=GNAI3%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=4387](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4387)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/2773>
- UniProt  
<http://www.uniprot.org/uniprot/P08754>

## **Sources for This Summary**

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